ABSTRACT

A genetically engineered mouse model that genotypically and phenotypically mimics human patients with spinal muscular atrophy. The genome of ~~said~~ the mouse model contains at least one mutation that knockouts the native mouse *Smn* gene and at least one copy of human *SMN^C^* gene that functions in a murine background and compensates for the loss of the functions provided by the *Smn* gene. The phenotypes of said mouse model can be grouped according to their severity of pathological conditions into three types, paralleling the three types of human spinal muscular atrophy conditions. Said mouse model can be used for studying the pathophysiology of spinal muscular atrophy and for developing and testing existing and new therapeutic and diagnostic methods.